Complete Summary

GUIDELINE TITLE

Health supervision for children with Down syndrome.

BIBLIOGRAPHIC SOURCE(S)

American Academy of Pediatrics, Section on Genetics. Health supervision for children with down syndrome. Pediatrics 2001 Feb;107(2):442-9. [23 references]

GUIDELINE STATUS

This is the current release of the guideline. This guideline updates a previously issued title "Health Supervision for Children with Down Syndrome" (Pediatrics 1994 May;93[5]:855-9).

AAP Policies are reviewed every 3 years by the authoring body, at which time a recommendation is made that the policy be retired, revised, or reaffirmed without change. Until the Board of Directors approves a revision or reaffirmation, or retires a statement, the current policy remains in effect.

COMPLETE SUMMARY CONTENT

SCOPE

DISEASE/CONDITION(S)

Down syndrome

GUIDELINE CATEGORY

Counseling
Diagnosis

Evaluation
Management

CLINICAL SPECIALTY

Family Practice
Medical Genetics
Obstetrics and Gynecology
Pediatrics

INTENDED USERS

Advanced Practice Nurses
Nurses
Physician Assistants
Physicians

GUIDELINE OBJECTIVE(S)

- To assist the pediatrician in caring for the child in whom the diagnosis of Down syndrome has been confirmed by karyotype
- To assist the pediatrician in counseling a pregnant women who has been given the prenatal diagnosis of Down syndrome

TARGET POPULATION

- Infants and children with Down syndrome
- Pregnant women given the prenatal diagnosis of Down syndrome

INTERVENTIONS AND PRACTICES CONSIDERED

Age-directed health supervision for children with Down syndrome and their parents including:

1. Prenatal and neonatal diagnostic assessment, including karyotype review, phenotype review, and discussion of recurrence risks
2. Anticipatory guidance, including early intervention services
3. Reproductive options, including family support, support groups, long-term planning, and sexuality
4. Medical evaluation, including growth measurements (height and weight), thyroid screening, hearing screening, vision screening, cervical spine roentgenogram, echocardiogram, and complete blood count (CBC)
5. Psychosocial issues, such as development and behavioral issues, school performance, and socialization
6. Assuring compliance with American Academy of Pediatrics recommendations for preventive pediatric health care

MAJOR OUTCOMES CONSIDERED

Not stated

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

Not stated

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Not stated

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Not applicable

METHODS USED TO ANALYZE THE EVIDENCE

Review

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Not stated

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Not applicable

COST ANALYSIS

A formal cost analysis was not performed and published cost analyses were not reviewed.

METHOD OF GUIDELINE VALIDATION

Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

Not stated

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

The Prenatal Visit

Pediatricians may be asked to counsel a family in which a fetus has a genetic disorder. In some settings, the pediatrician may be the primary resource for counseling. At other times, counseling may have been provided for the family by a clinical geneticist, obstetrician, or developmental pediatrician. In addition, parents may have received information from a Down syndrome program, a national Down syndrome organization, or an Internet site. Because of a previous relationship with the family, the pediatrician may be asked to review this information and assist in the decision-making process. As appropriate, the pediatrician should discuss the following topics with the family:

1. The prenatal laboratory or fetal imaging studies leading to the diagnosis.
2. The mechanism for occurrence of the disorder in the fetus and the potential recurrence rate for the family.
3. The prognosis and manifestations, including the wide range of variability seen in infants and children with Down syndrome.
4. When applicable, additional studies that may refine the estimation of the prognosis (e.g., fetal echocardiogram, ultrasound examination for gastrointestinal malformations).
5. Currently available treatments and interventions. This discussion needs to include the efficacy, potential complications and adverse effects, costs, and other burdens associated with these treatments. Discuss early intervention resources, parent support programs, and any plausible future treatments.
6. The options available to the family for management and rearing of the child using a nondirective approach. In cases of early prenatal diagnosis, this may include discussion of pregnancy continuation or termination, rearing the child at home, foster care placement, and adoption.

If the pregnancy is continued, a plan for delivery and neonatal care must be developed with the obstetrician and the family. Offer parent-to-parent contact. As the pregnancy progresses, additional studies may be valuable for modifying this management plan (e.g., detection of a complex heart defect by echocardiography). When appropriate, referral to a clinical geneticist should be considered for a more extended discussion of clinical outcomes and variability, recurrence rates, future reproductive options, and evaluation of the risks for other family members.

Health Supervision from Birth to 1 Month: Newborns

Examination

Confirm the diagnosis of Down syndrome and review the karyotype with the parents. Review the phenotype. Discuss the specific findings with both parents whenever possible, and talk about the following potential clinical manifestations

associated with the syndrome. These may have to be reviewed again at a subsequent meeting.

Discuss and Review

- Hypotonia
- Facial appearance

Evaluate for

- Feeding problems
- Strabismus, cataracts, and nystagmus at birth or by 6 months
- Congenital hearing loss with objective testing, such as brainstem auditory evoked response or otoacoustic emission at birth or by 3 months (Dahle & McCollister, 1986)
- Heart defects (approximately 50% risk). Perform cardiac evaluation (consultation by a cardiologist with expertise and experience in pediatric patients and echocardiogram recommended).
- Duodenal atresia
- Constipation with increased risk of Hirschsprung disease
- Leukemia–more common in children with Down syndrome than in the general population, but still rare (<1%); leukemoid reactions, on the other hand, are relatively common as is polycythemia (18%). Obtain complete blood count.
- Congenital hypothyroidism (1% risk)
- Respiratory tract infections

Anticipatory Guidance

- Discuss increased susceptibility to respiratory tract infection.
- Discuss the availability and efficacy of early intervention.
- Discuss the early intervention services in the community.
- Inform the family of the availability of support and advice from the parents of other children with Down syndrome.
- Supply names of Down syndrome support groups and current books and pamphlets (see "Bibliography and Resources for New Parents" in the original guideline document).
- Discuss the strengths of the child and positive family experiences.
- Check on individual resources for support, such as family, clergy, and friends.
- Talk about how and what to tell other family members and friends. Review methods of coping with long-term disabilities.
- Review the recurrence risk in subsequent pregnancies and the availability of prenatal diagnosis.
- Discuss unproven therapies. (National Down Syndrome Congress, 1989, 1999; Nickel, 1996; National Down Syndrome Society, 1997; National Down Syndrome Society, 1996; Spigelblatt et al., 1994; American College of Medical Genetics, 1996)

Health Supervision from 1 Month to 1 Year: Infancy

Examination

Physical Examination and Laboratory Studies

- Review the risk of serous otitis media (50%-70%). If the tympanic membranes cannot be visualized, or if the parents express any concern about their child's hearing, refer the infant to an otolaryngologist. Review the prior hearing evaluation (brainstem auditory evoked response and otoacoustic emission) and refer back to the otolaryngologist and audiologist if the initial evaluation was abnormal for follow up examination and testing. A behavioral audiogram should be obtained at 1 year in all children examined.
- Check for strabismus, cataracts, and nystagmus by 6 months, if not done at birth. Check the infant's vision at each visit, using developmentally appropriate subjective and objective criteria. By 6 months, refer the infant to a pediatric ophthalmologist or an ophthalmologist with special expertise and experience with infants with disabilities.
- Verify results of newborn thyroid function screen. Because of increased risk of acquired thyroid disease, repeat at 6 and 12 months and then annually. (Cutler, Benezra-Obeiter, & Brink, 1986; Karlsson et al., 1998)
- Administer pneumococcal vaccine, as well as other vaccines recommended for all children unless there are specific contraindications.

Anticipatory Guidance

- Review the infant's growth and development relative to other children with Down syndrome (see Figures 1-4 in the original guideline document) (Cronk et al., 1988)
- Review availability of Down syndrome support groups (see "Bibliography and Resources for New Parents" in the original guideline document).
- Assess the emotional status of parents and intrafamily relationships. Educate and support siblings and discuss sibling adjustments. At 6 to 12 months, review the psychological support and intrafamily relationships, including long-term planning, financial planning, and guardianship.
- Review the early intervention services relative to the strengths and needs of the infant and family (see "Bibliography and Resources for New Parents" in the original guideline document).
- Review the family's understanding of the risk of recurrence of Down syndrome and the availability of prenatal diagnosis.

Health Supervision from 1 to 5 Years: Early Childhood

- Obtain a history and perform a physical examination with attention to growth and developmental status.
- Review the risk of serous otitis media with hearing loss. If the tympanic membranes cannot be completely visualized (because of the frequent problem of stenotic ear canals), check the child's audiogram every 6 months up to 3 years or up to when a pure tone audiogram is obtained. Refer the child to an otolaryngologist or audiologist if necessary (approximately 50%-70% risk of serous otitis media between 3 and 5 years).
- Check the child's vision annually, using developmentally appropriate subjective and objective criteria. Refer the child to a pediatric ophthalmologist or ophthalmologist with special expertise and experience with children with disabilities every 2 years (approximately 50% risk of refractive errors between 3 and 5 years).

- At 3 to 5 years, obtain radiographs for evidence of atlantoaxial instability or subluxation. These may be obtained once during the preschool years. The need for these studies has been questioned, but they may be required for participation in the Special Olympics. These studies are more important for children who may participate in contact sports and are indicated in those who are symptomatic. (Davidson, 1988; Msall et al., 1990; Pueschel et al., 1987; Pueschel & Scola, 1987)
- Perform thyroid screening tests annually.
- Discuss symptoms related to obstructive sleep apnea, including snoring, restless sleep, and sleep position. Refer to a specialist as indicated. (Pueschel & Pueschel, 1992)

Anticipatory Guidance

- Review early intervention, including physical therapy, occupational therapy, and speech, in the preschool program and discuss future school placement and performance.
- Discuss future pregnancy planning, risk of recurrence of Down syndrome, and prenatal diagnosis.
- Assess the child's behavior, and talk about behavioral management, sibling adjustments, socialization, and recreational skills.
- Encourage families to establish optimal dietary and physical exercise patterns that will prevent obesity.

Health Supervision from 5 to 13 Years: Late Childhood

- Obtain a history and perform a physical examination with attention to growth and developmental status.
- Obtain audiologic evaluation annually.
- Obtain ophthalmologic evaluation annually.
- Perform thyroid screening tests annually (3%-5% risk of hypothyroidism).
- If appropriate, discuss skin problems: very dry skin and other skin problems are particularly common in patients with Down syndrome.
- Discuss symptoms related to obstructive sleep apnea, including snoring, restless sleep, and sleep position. Refer to a specialist as indicated. (Pueschel & Pueschel, 1992)

Anticipatory Guidance

- Review the child's development and appropriateness of school placement and developmental intervention.
- Discuss socialization, family status, and relationships, including financial arrangements and guardianship.
- Discuss the development of age-appropriate social skills, self-help skills, and the development of a sense of responsibility.
- Discuss psychosexual development, physical and sexual development, menstrual hygiene and management, fertility, and contraception. (de la Cruz & LaVeck, 1973)
- Discuss the need for gynecologic care in the pubescent female. Talk about the recurrence risk of Down syndrome with the patient and her family if she were to become pregnant. (Jagiello, 1981) Review the fact that although there

have been 2 case reports in which a male has reproduced, males with Down syndrome are usually infertile. (Jagiello, 1981)

Health Supervision from 13 to 21 Years or Older: Adolescence to Early Adulthood

Examination

- Perform physical examination including complete blood count (CBC) and thyroid function tests.
- Obtain annual audiologic evaluation.
- Obtain annual ophthalmologic evaluation.
- Discuss skin care.

Anticipatory Guidance

- Discuss issues related to transition into adulthood.
- Discuss appropriateness of school placement with emphasis on adequate vocational training within the school curriculum. (Pueschel & Pueschel, 1992; Fenner, Hewitt, and Torpy, 1987)
- Talk about the recurrence risk of Down syndrome with the patient and her family if she were to become pregnant. (Jagiello, 1981)
- Discuss sexuality and socialization. Discuss the need for and degree of supervision and/or the need for contraception. Make recommendations for routine gynecologic care.
- Discuss group homes and independent living opportunities, workshop settings, and other community-supported employment.
- Discuss intrafamily relationships, financial planning, and guardianship.
- Facilitate transfer to adult medical care.

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

REFERENCES SUPPORTING THE RECOMMENDATIONS

References open in a new window

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of evidence supporting each recommendation is not specifically stated.

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

Medical management, home environment, education, and vocational training can significantly affect the level of functioning of children and adolescents with Down syndrome and facilitate their transition to adulthood.

POTENTIAL HARMS

Not stated

QUALIFYING STATEMENTS

QUALIFYING STATEMENTS

The recommendations in this policy statement do not indicate an exclusive course of treatment for children with genetic disorders, but are meant to supplement anticipatory guidelines available for treating the healthy child provided in the American Academy of Pediatrics publication, "Guidelines for Health Supervision." They are intended to assist the pediatrician in helping children with genetic conditions to participate fully in life. Diagnosis and treatment of genetic disorders are changing rapidly. Therefore, pediatricians are encouraged to view these guidelines in the light of evolving scientific information. Clinical geneticists may be a valuable resource for the pediatrician seeking additional information or consultation.

The recommendations in this statement do not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Living with Illness

IOM DOMAIN

Effectiveness
Patient-centeredness

IDENTIFYING INFORMATION AND AVAILABILITY

BIBLIOGRAPHIC SOURCE(S)

American Academy of Pediatrics, Section on Genetics. Health supervision for children with down syndrome. Pediatrics 2001 Feb;107(2):442-9. [23 references]

ADAPTATION

Not applicable: The guideline was not adapted from another source.

DATE RELEASED

2001 Feb

GUIDELINE DEVELOPER(S)

American Academy of Pediatrics - Medical Specialty Society

SOURCE(S) OF FUNDING

American Academy of Pediatrics

GUIDELINE COMMITTEE

Committee on Genetics

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Committee on Genetics, 2000-2001: Christopher Cunniff, MD, Chairperson; Jaime L. Frias, MD; Celia Kaye, MD, PhD; John B. Moeschler, MD; Susan R. Panny, MD; Tracy L. Trotter, MD

Liaison Representatives: Felix de la Cruz, MD, MPH (National Institute of Child Health and Human Development); John Williams III, MD (American College of Obstetricians and Gynecologists); James W. Hanson, MD (American College of Medical Genetics); Cynthia A. Moore, MD, PhD (Centers for Disease Control and Prevention); Michele Lloyd-Puryear, MD, PhD (Health Resources and Services Administration)

Section Representative: H. Eugene Hoyme, MD (Section on Genetics)

Consultants: Rebecca S. Wappner, MD

Staff: Lauri Hall

FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

GUIDELINE STATUS

This is the current release of the guideline. This guideline updates a previously issued title "Health Supervision for Children with Down Syndrome" (Pediatrics 1994 May;93[5]:855-9).

AAP Policies are reviewed every 3 years by the authoring body, at which time a recommendation is made that the policy be retired, revised, or reaffirmed without change. Until the Board of Directors approves a revision or reaffirmation, or retires a statement, the current policy remains in effect.

GUIDELINE AVAILABILITY

Electronic copies: Available from the American Academy of Pediatrics (AAP) Policy Web site.

Print copies: Available from American Academy of Pediatrics, 141 Northwest Point Blvd., P.O. Box 927, Elk Grove Village, IL 60009-0927.

AVAILABILITY OF COMPANION DOCUMENTS

None available

PATIENT RESOURCES

None available

NGC STATUS

This summary was completed by ECRI on October 17, 2001. The information was verified by the guideline developer as of December 5, 2001.

COPYRIGHT STATEMENT

This NGC summary is based on the original guideline, which is subject to the guideline developer's copyright restrictions. Please contact the Permissions Editor, American Academy of Pediatrics (AAP), 141 Northwest Point Blvd, Elk Grove Village, IL 60007.

DISCLAIMER

NGC DISCLAIMER

The National Guideline Clearinghouse™ (NGC) does not develop, produce, approve, or endorse the guidelines represented on this site.

All guidelines summarized by NGC and hosted on our site are produced under the auspices of medical specialty societies, relevant professional associations, public or private organizations, other government agencies, health care organizations or plans, and similar entities.

Guidelines represented on the NGC Web site are submitted by guideline developers, and are screened solely to determine that they meet the NGC Inclusion Criteria which may be found at http://www.guideline.gov/about/inclusion.aspx.

NGC, AHRQ, and its contractor ECRI make no warranties concerning the content or clinical efficacy or effectiveness of the clinical practice guidelines and related materials represented on this site. Moreover, the views and opinions of developers or authors of guidelines represented on this site do not necessarily state or reflect those of NGC, AHRQ, or its contractor ECRI, and inclusion or hosting of guidelines in NGC may not be used for advertising or commercial endorsement purposes.

Readers with questions regarding guideline content are directed to contact the guideline developer.

© 1998-2006 National Guideline Clearinghouse

Date Modified: 9/25/2006